

# ALPHA-1

## ***FAST FACTS***

Alpha-1 Antitrypsin Deficiency (Alpha-1) is a genetic condition - passed on from parents to their children through genes. Alpha-1 may result in serious lung disease in adults and/or liver disease at any age. In the United States there are at least 100,000 people with Alpha-1 (ZZ).

### **COMMON SIGNS & SYMPTOMS OF ALPHA-1:**

- Shortness of breath, particularly with exertion
- Wheezing
- Chronic cough and sputum (phlegm) production (chronic bronchitis)
- Recurring chest colds
- Decreased exercise tolerance
- Bronchiectasis
- Unexplained liver disease or elevated liver enzymes
- Eyes and skin turning yellow (jaundice)
- Swelling of the abdomen (ascites)

Alpha-1 has been identified in virtually all populations. Worldwide, the number of people with Alpha-1 is 1 in every 1,500 to 3,500 people. An estimated 19 million people in the U.S. have one normal and one defective Alpha-1 gene.



People with one normal Alpha-1 gene (M) and one defective Alpha-1 gene (usually Z or S) are called "carriers". Carriers may have lower blood levels of Alpha-1 protein and can pass the defective gene to their children.

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Alpha-1 can lead to lung destruction and is often misdiagnosed with more common conditions like asthma or smoking-related Chronic Obstructive Pulmonary Disease (COPD).



Alpha-1 is the most common known genetic risk factor for emphysema and COPD. About 3% of all people diagnosed with COPD have undetected Alpha-1.

The American Thoracic Society (ATS) and the European Respiratory Society (ERS) recommend that everyone diagnosed with COPD, emphysema, or asthma that is not completely reversible with aggressive treatment be tested for Alpha-1.



Alpha-1 can cause liver problems in infants, children, and adults. Some people with Alpha-1 will have little or no liver disease, while others may develop severe liver disease. The most serious liver diseases are cirrhosis and liver cancer.

Alpha-1 cannot be diagnosed by symptoms or by a medical examination alone; this simple blood test is the only way doctors can diagnose the condition.

